

Chapter 2

Predictive Genetic Testing

The second chapter gives an introduction to and analysis of predictive genetic testing (PGT). This chapter explains PGT to identify the distinguishing characteristics of PGT that shape the revised model of consent in future chapters.

In order to fully understand the distinguishing characteristics, the beginning of the chapter will focus on the science of PGT. Having an understanding of the science behind this testing will help to elicit and further develop the three distinctive characteristics for this and future chapters. This chapter will introduce each of the three characteristics that arise from the science of PGT. As a result, this chapter establishes the foundation of the analysis for all the subsequent chapters.

The first part of Chap. 2 focuses on the background and science of PGT in order to get a better idea of the technical aspects behind genetic testing. The second part deals with the first distinguishing characteristic of PGT, understanding genetic risks and probabilities. Because genetic risks and probabilities are typically unique to PGT, this section will look at possible misunderstandings that can arise from not fully understanding genetic information. The third part looks at treatment options for diagnosed genetic traits. PGT can be different from other medical tests in that this testing can return results for diseases that have no treatment or preventative measures. This section will analyze that fact and identify some possible treatments that might help with PGT. The fourth area analyzes family-related genetic information. While some medical tests can involve family information, PGT has significant implications for family members of the individuals being tested. This section will discuss different styles of communication and disclosure of test results, and it will also look at the different concerns of family-related information for genetics. Then this fourth area will analyze the ideas of genetic exceptionalism versus normal medical information in regards to family-related genetic information. The fifth section will conclude with a summary of the implications and the distinguishing characteristics of PGT. By identifying the distinguishing characteristics of genetics, the chapter will highlight the power and limitations of genetic information and testing. The conclusion will bring together all the aspects of PGT that can influence the revised model of informed consent.

A. The Science Behind PGT

This section will look at PGT and the science behind it including issues of new genetic technologies and genetic information associated with specific diseases. Predictive genetic testing (PGT) looks at those at risk, the asymptomatic people.¹ One of the differences between PGT and a typical medical diagnostic test is the fact that PGT looks at the future while a diagnostic test gives information concerning the present.²

An article by Philip Mitchell, Bettina Meiser, Alex Wilde, et al. defines genetic testing as a test “used to identify a particular genotype (or set of genotypes) for a particular disease in a particular population for a particular purpose.”³ Philip Mitchell, Bettina Meiser, Alex Wilde, et al. go on to say that population is important because of the positive predictive value (PPV). The predictive value of a test is influenced by how often that disease occurs in a specific population. Generally there are three concepts used to evaluate a test: analytical validity, clinical validity, and clinical utility. The article by Philip Mitchell, Bettina Meiser, Alex Wilde, et al. says that analytical validity is related to the reliability and accuracy of a specific test. In genetic testing this is the ability of the test to identify the specific genotype. Analytical validity answers the question of does the genetic test actually identify what it is supposed to identify such as a Huntington’s mutation or the BRCA mutation. Clinical validity is “determined by: (1) the strength of evidence for the link between genotype and disease; and (2) test performance characteristics such as sensitivity, specificity, positive and negative predictive values, and likelihood ratios.”⁴ Clinical validity looks at the accuracy and consistency of test performance. Clinical utility looks at the actual value of the test. This area answers the question of whether or not the test provides information that could be useful to the patient. There are 8 areas that should be analyzed in clinical utility. The testing purpose should look at areas of legitimacy, efficacy, effectiveness, and appropriateness. The possibility of testing should analyze areas of acceptability, efficiency of economic evaluation, optimality of economic evaluation, and equity of resources.⁵

This science behind PGT will look at four different areas including the science, utility, benefits, and risks of testing. The science will look at the technical aspects of testing, the utility will provide the basic value and purpose of testing, and then the benefits and risks of testing will be analyzed.

¹ Anita Silvers and Michael Ashley Stein, “An Equality Paradigm for Preventing Genetic Discrimination,” 1342; Michael J. Green and Jeffrey R. Botkin, ““Genetic Exceptionalism” in Medicine: Clarifying the Differences between Genetic and Nongenetic Tests,” 571.

² James Evans, Cecile Skrzynia, and Wylie Burke, “The Complexities of Predictive Genetic Testing,” 1053.

³ Philip Mitchell, Bettina Meiser, Alex Wilde, et al., “Predictive and Diagnostic Genetic Testing in Psychiatry,” 227–228.

⁴ Philip Mitchell, Bettina Meiser, Alex Wilde, et al., “Predictive and Diagnostic Genetic Testing in Psychiatry,” 227–228.

⁵ Philip Mitchell, Bettina Meiser, Alex Wilde, et al., “Predictive and Diagnostic Genetic Testing in Psychiatry,” 227–228.

1. Science

The purpose of PGT is to assess an individual's risk of developing a specific disease. PGT analyzes DNA and genetics to identify whether a person will develop or already has an at-risk gene that causes them to have a higher or lower than an average risk of developing a specific disease. In order to make predictions about a person's risk for developing a specific disease, scientists analyze areas in a person's genes and chromosomes for genetic variants and mutations.⁶ PGT looks at "polymorphisms that increase the probability of disease development."⁷ Mutations can be identified by looking at segments of genes and chromosomes in order to identify genes that are different from the normal size and shape of a specific gene.⁸ Currently, studies have identified the genetic variants that might be linked to a higher risk of at least 40 diseases.⁹ Also studies have identified over 1000 genetic variants associated with a risk of disease.¹⁰ The genetic variants make up what is called a single nucleotide polymorphism (SNP). Generally companies predict genetic risks by "calculating how often that condition occurs among people of the customer's general age, sex and ethnicity, then factor in the presence or absence of the relevant SNP."¹¹ It is important to note that the results depend on what genetic variants the laboratory decides to use when analyzing the predicted risk of disease.¹² One company could be analyzing 9 variants, while the other company is analyzing 13 variants. The number of variants analyzed can make a difference in the results as well. Once a genetic variant is associated with a risk of a particular disease, tests can be run to look at the SNPs in a specific sample in order to determine risk. If the individual does have the mutation, then it means that that individual has a higher chance of developing that specific disease or cancer than a person without that mutation.¹³

Diseases can be caused by a number of genetic and environmental factors. An important aspect to predicting disease is the fact that there can be several variants in a gene and those variants can be linked to several different diseases.¹⁴ SNPs can be associated with many different areas. One SNP could be more common in a person

⁶ Anita Silvers and Michael Ashley Stein, "An Equality Paradigm for Preventing Genetic Discrimination," 1346–1347.

⁷ Michael J. Green and Jeffrey R. Botkin, "Genetic Exceptionalism" in Medicine: Clarifying the Differences between Genetic and Nongenetic Tests," 571.

⁸ Abigail L. Rose, Nikki Peters, Judy A. Shea, et al., "Attitudes and Misconceptions about Predictive Genetic Testing for Cancer Risk," 146.

⁹ Peter Kraft, Ph.D. and David Hunter, "Genetic Risk Prediction—Are We There Yet?" 1701.

¹⁰ Pauline C. Ng, Sarah S. Murray, Samuel Levy, et al., "An Agenda for Personalized Medicine," 724–725.

¹¹ Chris Berdik, "Genetic Tests Give Consumers Hints About Disease Risk; Critics Have Misgivings," 2.

¹² Francis Collins, *The Language of Life*, xxi–xxii.

¹³ Abigail L. Rose, Nikki Peters, Judy A. Shea, et al., "Attitudes and Misconceptions about Predictive Genetic Testing for Cancer Risk," 146.

¹⁴ Anita Silvers and Michael Ashley Stein, "An Equality Paradigm for Preventing Genetic Discrimination," 1346, 1385.

with breast cancer, Parkinson's disease, or another type of cancer.¹⁵ The BRCA 1/2 gene mutations were identified in 1994 and 1995.¹⁶ These mutations increase the likelihood of developing breast and/or ovarian cancer. Generally many different genes are involved in the development of a disease. For example, Anita Silvers and Michael Ashley Stein in the *Vanderbilt Law Review* say that researchers have found over eight hundred gene mutations that are linked to cystic fibrosis. Since there are so many different variations and mutations for this disease, the predictability of developing cystic fibrosis can vary greatly from variation to variation. One genetic mutation can impact the severity of the disease while another mutation might not impact the disease at all. Anita Silvers and Michael Ashley Stein in the *Vanderbilt Law Review* state that "identical mutations in such genes will affect individuals from different populations to different degrees because of variations in environmental factors."¹⁷ Each mutation does not carry the same weight.

Huntington's disease is more of a unique disease for PGT in that the test has a higher degree of certainty for predicting. In 1993, the genetic variation for Huntington's disease was identified, and as a result PGT was offered for at-risk individuals. Huntington's disease is an incurable neurodegenerative disease with no medical treatments available to slow the progression.¹⁸ There are approximately 30,000 individuals with Huntington's disease, but it is suggested that there are another 200,000 that have not been tested yet and are at risk in the United States. This disease is generally categorized as a late-onset condition, because typically the disease manifests itself around 40 years old. The test for Huntington's disease has 100% penetrance, which means if the gene is present, then the individual will develop Huntington's disease at some point in his or her life. On the other side, if the gene is not present, then the individual is not at risk for Huntington's disease. With this disease, either the Huntington's gene is present or it is absent. Because of the certainty and the predictive value of testing for Huntington's disease, the results for PGT are more clear cut. The science behind PGT for Huntington's disease is not as complicated as other diseases. However, there are other issues to consider with this testing such as the non-existent treatment options and the difficult family implications of this predictive testing, and both of these issues will be discussed in later sections.¹⁹

The confidence in predictive genetic testing can be both over- and underestimated at times. Most genetic tests will not predict with certainty the likelihood of develop-

¹⁵ Chris Berdik, "Genetic Tests Give Consumers Hints About Disease Risk; Critics Have Misgivings," 2; Marion Harris, Ingrid Winship, and Merle Spriggs, "Controversies and Ethical Issues in Cancer-Genetics Clinics," 301.

¹⁶ Abigail L. Rose, Nikki Peters, and Judy A. Shea, et al., "Attitudes and Misconceptions about Predictive Genetic Testing for Cancer Risk," 145.

¹⁷ Anita Silvers and Michael Ashley Stein, "An Equality Paradigm for Preventing Genetic Discrimination," 1385.

¹⁸ Susan M. Cox, "Stories in Decisions: How At-Risk Individuals Decide to Request Predictive Testing for Huntington Disease," 258.

¹⁹ Kathryn Holt, "What Do We Tell the Children? Contrasting the Disclosure Choices of Two HD Families Regarding Risk Status and Predictive Genetic Testing," 254-255.

ing a specific disease, because most diseases have a number of genetic and environmental aspects to them. Huntington’s disease, however, has a genetic test that will predict with a high degree of certainty that a person will develop the disease in the future. But even with a disease like Huntington’s, there is no way to predict how the disease will affect an individual person. Many of the tests are subject to uncertainty, false positives or negatives, and possible misinterpretation. Anita Silvers and Michael Ashley Stein in the *Vanderbilt Law Review* say “neither now nor in the future will someone’s genetic makeup forecast that person’s future health condition with certainty.”²⁰ There are many factors that can influence the development and severity of the disease or illness. Some of the factors that influence the predictive value of the test include the differences of gene expression, accuracy of the specific test, and reliability of the research. But on the other side, Anita Silvers and Michael Ashley Stein in the *Vanderbilt Law Review* say “it is equally misleading to say that basing health predictions on genetic testing is ‘little more than medical speculation.’”²¹ So while PGT cannot attest to the severity and/or certainty of a specific disease, this testing does have a certain value and legitimacy for medical care.²²

Also included within the science of PGT are the inheritance patterns for disease. If one parent is homozygous for a specific disease, then he or she has two copies of the mutation. If a parent is heterozygous for a disease, then he or she has one copy of the mutation and one copy of the normal gene. Because there are different patterns of inheritance, not all disease inheritance is the same. For example, Huntington’s disease is an autosomal dominant disease. The inheritance pattern for Huntington’s disease is easy, because it is only concerned with one gene. Because it is dominant, the disease will be passed down to the children if any of the children inherit one mutated gene. The Punnett square below further illustrates this concept. The capital “H” is the mutation causing Huntington’s disease, while the lower case “h” represents the normal gene (Table 2.1).

The box represents one parent with Huntington’s disease (Hh in bold) and one parent without Huntington’s disease (hh in *italic*). When children are born, each child will have a 50 % chance of inheriting Huntington’s disease (Hh) and a 50 % chance of not having Huntington’s disease (hh). This is a basic representation of genetic inheritance patterns. While this seems fairly easy, the inheritance pattern for

Table 2.1 Punnett square for Huntington’s disease

	H	h
h	Hh	hh
h	Hh	hh

²⁰ Anita Silvers and Michael Ashley Stein, “An Equality Paradigm for Preventing Genetic Discrimination,” 1347–1348.

²¹ Anita Silvers and Michael Ashley Stein, “An Equality Paradigm for Preventing Genetic Discrimination,” 1347–1348.

²² Susan Wolf and Jeffrey Kahn, “Genetic Testing and the Future of Disability Insurance: Ethics, Law & Policy,” 8; Anita Silvers and Michael Ashley Stein, “An Equality Paradigm for Preventing Genetic Discrimination,” 1347–1348.

Huntington's disease is probably one of the easiest. Most other diseases that can be predicted with PGT are multifactorial diseases. There are several factors that influence the inheritance of the BRCA mutation. Even if a person has the mutation, it is not necessarily a positive diagnosis since there are other influencing factors such as the environment and the individual's lifestyle.²³

2. *Utility*

Sometimes PGT is recommended based on the utility of the testing, and other times it is not recommended because of a lack of insufficient evidence of testing benefit. Some instances of increased utility include the following: "high morbidity and mortality of disease, effective but imperfect treatment, high predictive power of the genetic test (high penetrance), high cost or onerous nature of screening and surveillance methods, and preventive measures that are expensive or associated with adverse effects."²⁴ On the other hand, decreased utility for predictive genetic testing include: "low morbidity and mortality of disease, highly effective and acceptable treatment, poor predictive power of the genetic test (low penetrance), availability of inexpensive, acceptable, and effective screening and surveillance methods, and preventive measures that are inexpensive, efficacious, and high acceptable—for example, vaccination."²⁵ James Evans, Cecile Skrzynia, and Wylie Burke in "The Complexities of Predictive Genetic Testing," from the *British Medical Journal* say that the usefulness of the test can decrease if the disease is curable. For example, James Evans, Cecile Skrzynia, and Wylie Burke suggest that when breast and colon cancer are able to be cured or treated by effective and safe measures, then the benefit of testing is reduced. However, if the disease is curable and is identified earlier, then the disease might be able to be cured earlier rather than later. Evans, Skrzynia, and Burke also suggest that if there are successful and economical screening tools in place for certain diseases, then the utility of PGT will decrease. One example given is of hypertension. Since there are acceptable screening methods that are not expensive, there is not a need to participate in PGT for hypertension. James Evans, Cecile Skrzynia, and Wylie Burke suggest that if the cost of screening is much higher, then PGT will be more economical and attractive to individuals. Evans, Skrzynia, and Burke also suggest that in order for PGT to have higher utility, the preventive measures would generally have some problems and/or be fairly costly. For example, the utility of PGT typically can increase when a person is at risk for breast cancer and is considering a prophylactic mastectomy. James Evans, Cecile

²³ Heidi Chial, "Mendelian Genetics: Patterns of Inheritance and Single-Gene Disorders;" Susan Wolf and Jeffrey Kahn, "Genetic Testing and the Future of Disability Insurance: Ethics, Law & Policy," 8.

²⁴ James Evans, Cecile Skrzynia, and Wylie Burke, "The Complexities of Predictive Genetic Testing," 1054.

²⁵ James Evans, Cecile Skrzynia, and Wylie Burke, "The Complexities of Predictive Genetic Testing," 1054–1055.

Skrzynia, and Wylie Burke say “when prevention is simple, however, the value of testing decreases,” and the example of a vaccination is given.²⁶ Since vaccinations are so easy to prevent diseases, there is no need for PGT of diseases like measles, mumps, and rubella. The utility of PGT can play an important role in the utilization of testing and the risks and benefits of testing.

3. *Benefits*

Reasons to undergo testing include motivational and emotional. Motivational reasons include “early detection, prevention, and control.”²⁷ The goal of PGT is to identify which people have the mutation so that additional monitoring can take place for those at risk. Identification and monitoring of at-risk individuals will hopefully “lead to reduced morbidity and mortality through targeted screening, surveillance, and prevention.”²⁸ If there is additional monitoring, then the hope is that there can be early diagnosis of the disease. PGT can help to monitor those at increased risk and decrease the amount of screening for those that are not at risk or are low risk. Possible prevention and treatment plans are another benefit of testing. Sometimes there can be surgeries or chemotherapy that can help to decrease a person’s risk for a specific disease.²⁹ Also PGT could help with future plans and “may lead individuals to alter their diet or avoid exposure to certain chemicals in an attempt to avoid future disease.”³⁰ The hope is that people will avoid “risk-inducing behaviors.”³¹ Shoshana Shiloh and Shiri Ilan, the authors of “To Test or Not To Test? Moderators of the Relationship Between Risk Perceptions and Interest in Predictive Genetic Testing,” described a study about risk perceptions and testing utilization. The study concluded that the high interest in the test was associated with “both motivations and especially with emotional-reassurance motivation, but not with risk perceptions, health/illness orientations, and cancer anxiety.”³² The study demonstrated that risk perceptions did not necessarily lead to increased test utilization or interest in PGT. Shoshana Shiloh and Shiri Ilan say that understanding the perceived risk is crucial but that is not enough to alter behaviors including fitness modifications. In order to change behaviors and goals, there needs to be psychological changes as well. The

²⁶ James Evans, Cecile Skrzynia, and Wylie Burke, “The Complexities of Predictive Genetic Testing,” 1054–1055.

²⁷ Clara L. Gaff, Veronica Collins, Tiffany Symes, et al., “Facilitating Family Communication about Predictive Genetic Testing: Probans’ Perceptions,” 133.

²⁸ James Evans, Cecile Skrzynia, and Wylie Burke, “The Complexities of Predictive Genetic Testing,” 1052.

²⁹ Neil Sharpe and Ronald Carter, *Genetic Testing: Care, Consent, and Liability*, 269–70.

³⁰ Susan Wolf and Jeffrey Kahn, “Genetic Testing and the Future of Disability Insurance: Ethics, Law & Policy,” 8.

³¹ Neil Sharpe and Ronald Carter, *Genetic Testing: Care, Consent, and Liability*, 269–70.

³² Shoshana Shiloh and Shiri Ilan, “To Test or Not To Test? Moderators of the Relationship Between Risk Perceptions and Interest in Predictive Genetic Testing,” 471.

article by Shoshana Shiloh and Shiri Ilan concludes that in order to have informed consent and decision making, there needs to be objective and reasonable information about the possible risks and benefits. Also there can be psychological and emotional benefits of testing. Emotional motivations include eliminating uncertainty, gaining support or hope, and preparing emotionally.³³ Finding out that a person is not at risk or is at a very low risk of developing a certain disease can often decrease his or her anxiety levels. This can also lead to a greater “self-perception.” Knowing a person’s risk status can help to alter or reinforce their view of themselves. Sometimes PGT can result in a greater “sense of control,” because the person at risk can follow certain procedures and/or treatments that could potentially decrease their risk.³⁴ In some people’s minds, PGT can help to gain control by knowing their risk status and/or organizing events in their life and future.³⁵ Motivational and emotional reasons for testing can be benefits of PGT.

There can also be future benefits. Anita Silvers and Michael Ashley Stein in the *Vanderbilt Law Review* suggest that some genetic variants and mutations could have different functions and outcomes than originally thought. “In the future, scientists could discover that having a particular breast cancer gene mutation correlates with immunity from AIDS (as sickle-cell trait correlates with heightened immunity to malaria).”³⁶ Even with emotional, motivational, and future benefits, sometimes there can also be anxiety, worry, and discrimination that can impact themselves and their family.³⁷

4. Risks

Shoshana Shiloh and Shiri Ilan in their article, “To Test or Not To Test? Moderators of the Relationship Between Risk Perceptions and Interest in Predictive Genetic Testing,” say that while PGT can promote encouragement and hope, it can also “cause considerable distress to others from premature knowledge of likely illness.”³⁸ Discrimination and psychological harms are often cited as the main harms, but there can be others associated with PGT. Sometimes there can be false assurances which can cause problems for future treatment. False assurances come from getting a lower risk than what is actually true. On the other hand, there can be problems from getting a higher risk than what is actually true. If a woman is told she

³³ Shoshana Shiloh and Shiri Ilan, “To Test or Not To Test? Moderators of the Relationship Between Risk Perceptions and Interest in Predictive Genetic Testing,” 471–472, 476–477.

³⁴ Neil Sharpe and Ronald Carter, *Genetic Testing: Care, Consent, and Liability*, 269–70.

³⁵ Abigail L. Rose, Nikki Peters, Judy A. Shea, et al., “Attitudes and Misconceptions about Predictive Genetic Testing for Cancer Risk,” 148.

³⁶ Anita Silvers and Michael Ashley Stein, “An Equality Paradigm for Preventing Genetic Discrimination,” 1385.

³⁷ Abigail L. Rose, Nikki Peters, Judy A. Shea, et al., “Attitudes and Misconceptions about Predictive Genetic Testing for Cancer Risk,” 148.

³⁸ Shoshana Shiloh and Shiri Ilan, “To Test or Not To Test? Moderators of the Relationship Between Risk Perceptions and Interest in Predictive Genetic Testing,” 469.

has a high risk for developing breast cancer, then she will make decisions based on that information such as having a prophylactic double mastectomy. Problems arise when people adopt “irreversible, risk-inducing or expensive risk prevention strategies based upon incorrectly high estimations of risk.”³⁹ Also sometimes people can have feelings of powerlessness. People often have no control over whether or not they develop a certain disease, and this can cause additional problems emotionally and even physically by not following recommended protocols. The possibility of discrimination and stigmatization can also be a risk of this information.⁴⁰

Discrimination can occur in many areas including employment, insurance, and social situations. Genetic discrimination “arises when individuals with no symptoms or signs receive less favorable or adverse treatment because of their genotype.”⁴¹ If the concern about discrimination is high, then sometimes people might not participate in PGT, because he or she is concerned about his family’s insurance premiums. If the tests are not conducted, then the individual could be missing out on possible treatment options. In a study with 163 cancer geneticists, about 68% of the geneticists said that if a person underwent testing for BRCA1 or 2 or hereditary non-polyposis colorectal cancer (HNPCC) they would not bill insurance so that there would be no discrimination. Also with this study, 26% of the geneticists said that they were in favor of using an alias for testing so as not to cause potential discrimination.⁴² Another study conducted by phone found that people were also discriminated against because a family member had a hereditary genetic disease. Anita Silvers and Michael Ashley Stein in the *Vanderbilt Law Review* say that having a negative test result could result in other prospects. Anita Silvers and Michael Ashley Stein continue to say that “Proof that they are not at risk will reassure them of their ability to succeed in endeavors aversive for people who develop the disease.”⁴³ Sometimes people who are at-risk will not participate in certain events or careers in life, because the individuals assume there is nothing he or she can do. Other times, people do not allow individuals to participate, because people consider those individuals to be at risk.⁴⁴ Discrimination can occur in many different areas and activities. At the beginning, individual and family discrimination played a significant part in PGT, but in 2003, the Genetic Information Non Discrimination Act (GINA) was established. This act tries to eliminate employment and insurance discrimination.⁴⁵ How-

³⁹ Neil Sharpe and Ronald Carter, *Genetic Testing: Care, Consent, and Liability*, 270.

⁴⁰ Douglas Martin and Heather Greenwood, “Public Perceptions of Ethical Issues Regarding Adult Predictive Genetic Testing,” 107.

⁴¹ Marion Harris, Ingrid Winship, Merle Spriggs, “Controversies and Ethical Issues in Cancer-Genetics Clinics,” 304–305.

⁴² Marion Harris, Ingrid Winship, Merle Spriggs, “Controversies and Ethical Issues in Cancer-Genetics Clinics,” 304–305.

⁴³ Anita Silvers and Michael Ashley Stein, “An Equality Paradigm for Preventing Genetic Discrimination,” 1349.

⁴⁴ Anita Silvers and Michael Ashley Stein, “An Equality Paradigm for Preventing Genetic Discrimination,” 1349.

⁴⁵ Marion Harris, Ingrid Winship, Merle Spriggs, “Controversies and Ethical Issues in Cancer-Genetics Clinics,” 305; Cynthia Marietta and Amy McGuire, “Direct-to-Consumer Genetic Testing: Is It the Practice of Medicine?” 370.

ever, Susan Wolf and Jeffrey Kahn in “Genetic Testing and the Future of Disability Insurance: Ethics, Law & Policy,” say that the “fear of discrimination is important, as individuals may decide to forego genetic testing (even when it might prove medically useful) in order to protect themselves against insurance discrimination.”⁴⁶

Another possible concern about knowing a person is at risk is psychological harms.⁴⁷ Marita Broadstock, Susan Michie, and Theresa Marteau in “Psychological Consequences of PGT: A Systematic Review” from the *European Journal of Human Genetics*, suggest that there were no significant changes in the emotional suffering for the carriers and non-carriers in a period of 3 years after PGT. The authors suggest a couple of reasons for this finding. In this study, there could be general psychological defense methods that had already been started. Broadstock, Michie, and Marteau say that research suggests that the people participating in testing are often stronger emotionally and are more capable of handling information. By already deciding to take a PGT and coming forward for testing, people tend to have thought about the testing in advance. Another study found that people getting tested for Huntington’s disease (HD) generally “had higher ego strength, were more socially extroverted, and had more positive coping strategies than the general population.”⁴⁸ Often the people considering getting tested for HD have increased experiential knowledge about this disease. Another study with HD looked at those who decided not to undergo testing and then compared them to those who came forward for testing. The study suggested that those who did not undergo HD testing were more pessimistic about the future. The most common attitudes presented in those who participated in the study were denial of the results or test, elimination of uncertainty, or both. Also sometimes people from at-risk families already had strong coping methods. Broadstock, Michie, and Marteau concluded that as genetic testing is increasingly brought into routine medical care, “some of the protective factors associated with the research environment are likely to be reduced,” and likely there will be more psychological harms.⁴⁹

However on the other hand, in “Predictive Genetic Testing in Children and Adults: A Study of Emotional Impact” from the *Journal of Medical Genetics*, S. Michie, M. Bobrow, and T. M. Marteau suggest that there is a significant level of anxiety after getting a positive test result. S. Michie, M. Bobrow, and T. M. Marteau say that this conclusion is especially important in adults with not as many psychological resources.⁵⁰ One article by Regina E. Ensenuer, Virginia V. Michels, and Shanda S. Reinke, “Genetic Testing: Practical, Ethical, and Counseling Consider-

⁴⁶ Susan Wolf and Jeffrey Kahn, “Genetic Testing and the Future of Disability Insurance: Ethics, Law & Policy,” 11.

⁴⁷ Neil Sharpe and Ronald Carter, *Genetic Testing: Care, Consent, and Liability*, 270.

⁴⁸ Marita Broadstock, Susan Michie, Theresa Marteau, “Psychological Consequences of Predictive Genetic Testing: A Systematic Review,” 735–736.

⁴⁹ Marita Broadstock, Susan Michie, Theresa Marteau, “Psychological Consequences of Predictive Genetic Testing: A Systematic Review,” 735–736.

⁵⁰ S. Michie, M. Bobrow, and T. M. Marteau, on behalf of the FAP Collaborative Research Group, “Predictive Genetic Testing in Children and Adults: A Study of Emotional Impact,” 520, 526.



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